

This listing of claims will replace all prior versions, and listings, of claims in the application:

Listing of Claims:

1-60. (cancelled)

61. (currently amended) A kit for assaying for the presence of a mutation associated with Familial Dysautonomia in an individual comprising primers 18F (SEQ ID NO:82) and 23R (SEQ ID NO:84) that are capable of amplifying a region of ~~IKAP~~ IKBKAP of sufficient size to detect a FD1 mutation or a FD2 mutation, wherein said region amplified comprises a FD1 or a FD2 mutation.

62-67. (cancelled)

68. (previously presented) The kit of claim 61, wherein the region amplified comprises position 2,397 of SEQ ID NO:2.

69-80. (cancelled)

81. (new) An oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, or the complement thereof, said oligonucleotide probe being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

82. (new) The oligonucleotide probe of claim 81 which is 16 nucleotides.

83. (new) An oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, or the complement thereof, said oligonucleotide probe being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1.

84. (new) The oligonucleotide probe of claim 83 which is 16 nucleotides.

85. (new) An oligonucleotide probe which consists of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ IN NO:1, or the complement thereof, said oligonucleotide probe being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

86. (new) An oligonucleotide probe which consists of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, or the complement thereof, said oligonucleotide probe being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1.

87. (new) An oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, or the complement thereof, except that the nucleotide corresponding to 34,201 is a cytosine, or a guanine in said complement, said oligonucleotide probe being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

88. (new) The oligonucleotide probe of claim 87 which is 16 nucleotides.

89. (new) An oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, or the complement thereof, except that the nucleotide corresponding to 33,714 is a cytosine, or a guanine in said complement, said oligonucleotide probe being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1.

90. (new) The oligonucleotide probe of claim 89 which is 16 nucleotides.

91. (new) An oligonucleotide probe which consists of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, or the complement thereof, except that the nucleotide corresponding to 34,201 is a cytosine, or a guanine in said complement, said oligonucleotide probe being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

92. (new) An oligonucleotide probe which consists of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, or the complement thereof, except that the nucleotide corresponding to 33,714 is a cytosine, or a guanine in said complement, said oligonucleotide probe being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

93. (new) A kit for the detection of a mutation associated with Familial Dysautonomia in a sample from a human subject, said kit comprising a pair of PCR primers that flank a mutation site in the IKBKAP gene corresponding to position 33,714 of SEQ ID NO:1 or position 34,201 of SEQ ID NO:1, said pair of PCR primers being capable of amplifying a portion of the IKBKAP gene containing position 33,714 or position 34,201 of SEQ ID NO:1.

94. (new) The kit of claim 93 wherein each of said PCR primers consists of at least 16 contiguous nucleotides of SEQ ID NO:1, or a complement thereof.

95. (new) The kit of claim 93 wherein each of said PCR primers consists of 16 contiguous nucleotides of SEQ ID NO:1, or a complement thereof.

96. (new) The kit of claim 93 wherein each of said PCR primers comprises 16 contiguous nucleotides of SEQ ID NO:1, or a complement thereof.

97. (new) The kit of claim 93, said portion of the IKBKAP gene being a portion from exon 18 through exon 23.

98. (new) The kit of claims 93 which further comprises an oligonucleotide probe selected from the group consisting of

(a) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1, which include position 34,201 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 34,201;

(b) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1, which include position 34,201 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 34,201;

(c) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(d) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(e) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, except that the nucleotide corresponding to 34,201 is a cytosine, and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(f) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1, which include position 34,201 of SEQ ID NO:1, except that the nucleotide corresponding to 34,201 in the complement is a guanine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(g) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, except that the nucleotide corresponding to 34,201 is a cytosine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(h) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1 except that the nucleotide corresponding to 34,201 in the complement is a guanine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

(i) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 and being suitable for the detection of the FD mutation at position 33,714;

(j) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 and being suitable for the detection of the FD mutation at position 33,714;

(k) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(l) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(m) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 except that the nucleotide corresponding to 33,714 is a cytosine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(n) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 except that the nucleotide corresponding to 33,714 in the complement is a guanine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(o) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which

include position 33,714 of SEQ ID NO:1, except that the nucleotide corresponding to 33,714 is a cytosine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1; and

(p) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, except that the nucleotide corresponding to 33,714 in the complement is a guanine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1.

99. (new) The oligonucleotide probe of claim 98 which is 16 nucleotides.

100. (new) A kit for the detection of a mutation associated with Familial Dysautonomia in a sample from a human subject, said kit comprising an oligonucleotide probe selected from the group consisting of

(a) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1 and being suitable for the detection of the FD mutation at position 34,201;

(b) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1 and being suitable for the detection of the FC mutation at position 34,201;

(c) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which

include position 34,201 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(d) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(e) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1 except that the nucleotide corresponding to 34,201 is a cytosine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(f) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1 except that the nucleotide corresponding to 34,201 in the complement is a guanine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(g) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID NO:1 which include position 34,201 of SEQ ID NO:1, except that the nucleotide corresponding to 34,201 is a cytosine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1;

(h) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 20 and intron 20 of the IKBKAP gene of SEQ ID

NO:1 which include position 34,201 of SEQ ID NO:1, except that the nucleotide corresponding to 34,201 in the complement is a guanine and being suitable for the detection of the FD mutation at position 34,201 of SEQ ID NO:1.

(i) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 and being suitable for the detection of the FD mutation at position 33,714;

(j) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 and being suitable for the detection of the FC mutation at position 33,714;

(k) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(l) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(m) an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 except that the nucleotide corresponding to 33,714 is a cytosine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(n) the complement of an oligonucleotide probe comprising 16 contiguous nucleotides of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1 except that the nucleotide corresponding to 33,714 in the complement is a guanine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1;

(o) an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 which include position 33,714 of SEQ ID NO:1, except that the nucleotide corresponding to 33,714 is a cytosine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1; and

(p) the complement of an oligonucleotide probe consisting of at least 16 contiguous nucleotides of the nucleotide sequence of exon 19 and intron 19 of the IKBKAP gene of SEQ ID NO:1 except that the nucleotide corresponding to 33,714 in the complement is a guanine and being suitable for the detection of the FD mutation at position 33,714 of SEQ ID NO:1.

101. (new) The oligonucleotide probe of claim 100 which is 16 nucleotides.